(12) INTERNATIONAL APPLICATION PUBLISHED UNDER THE PATENT COOPERATION TREATY (PCT)

(19) World Intellectual Property Organization
International Bureau

(43) International Publication Date
2 February 2006 (02.02.2006)

PCT

(10) International Publication Number
WO 2006/011716 A1

(51) International Patent Classification[7]: C12Q 1/68

(21) International Application Number:
PCT/KR2005/002170

(22) International Filing Date: 6 July 2005 (06.07.2005)

(25) Filing Language: Korean

(26) Publication Language: English

(30) Priority Data:
10-2004-0052652 7 July 2004 (07.07.2004) KR

(71) Applicant *(for all designated States except US)*: **KONGJU NATIONAL UNIVERSITY INDUSTRY ACADEMIA COOPERATION GROUP** [KR/KR]; Sanhak Research Hall 104-ho, Kongju, National University, 182 Sinkwan-dong, Kongju-si, Chungcheongnam-do, 314-701 (KR).

(71) Applicant and

(72) Inventor: **CHOI, Byung Ok** [KR/KR]; 6-104 Sunkyung Apt., 506 Daechi-dong, Gangnam-gu, Seoul, 135-836 (KR).

(72) Inventor; and

(75) Inventor/Applicant *(for US only)*: **CHUNG, Ki Wha** [KR/KR]; 214-606 Sameori Apt., 908 Dunsan-dong, Seo-gu, Daejeon, 302-777 (KR).

(74) Agent: **YOU, Byung Sun**; 610 Mannyun Officetel, 241 Walpyung-dong, Seo-gu, Daejeon, 302-282 (KR).

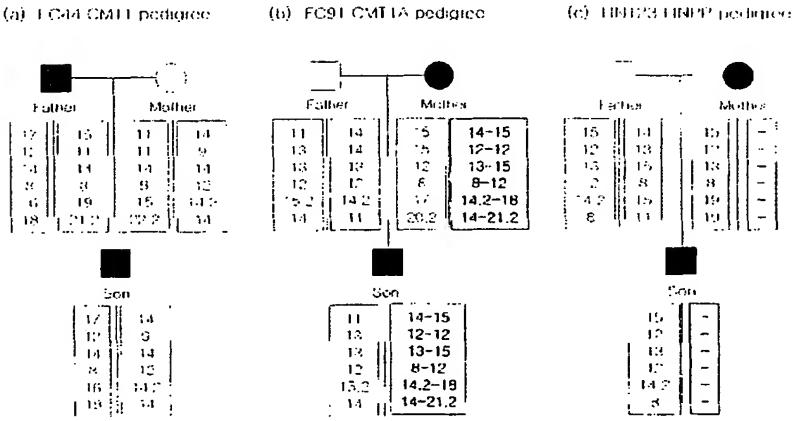
(81) Designated States *(unless otherwise indicated, for every kind of national protection available)*: AE, AG, AL, AM, AT, AU, AZ, BA, BB, BG, BR, BW, BY, BZ, CA, CH, CN, CO, CR, CU, CZ, DE, DK, DM, DZ, EC, EE, EG, ES, FI, GB, GD, GE, GH, GM, HR, HU, ID, IL, IN, IS, JP, KE, KG, KM, KP, KZ, LC, LK, LR, LS, LT, LU, LV, MA, MD, MG, MK, MN, MW, MX, MZ, NA, NG, NI, NO, NZ, OM, PG, PH, PL, PT, RO, RU, SC, SD, SE, SG, SK, SL, SM, SY, TJ, TM, TN, TR, TT, TZ, UA, UG, US, UZ, VC, VN, YU, ZA, ZM, ZW.

(84) Designated States *(unless otherwise indicated, for every kind of regional protection available)*: ARIPO (BW, GH, GM, KE, LS, MW, MZ, NA, SD, SL, SZ, TZ, UG, ZM, ZW), Eurasian (AM, AZ, BY, KG, KZ, MD, RU, TJ, TM), European (AT, BE, BG, CH, CY, CZ, DE, DK, EE, ES, FI, FR, GB, GR, HU, IE, IS, IT, LT, LU, LV, MC, NL, PL, PT, RO, SE, SI, SK, TR), OAPI (BF, BJ, CF, CG, CI, CM, GA, GN, GQ, GW, ML, MR, NE, SN, TD, TG).

Published:
— *with international search report*

[Continued on next page]

(54) Title: DIAGNOSIS METHOD AND KITS FOR INHERITED NEUROPATHIES CAUSED BY DUPLICATION OR DELETION OF CHROMOSOME 17P11.2-P12 REGION



(57) Abstract: Disclosed herein are a method and kit for diagnosing hereditary diseases CMT1A and HNPP, caused by duplicatino and deletion in the chromosome 17pl 1.2-pl2 region. In accordance with the present invention, there is provided a method for diagnosing an inherited neuropathy, comprising, runnung the PCR amplification using microsatellites present in a chromosome 17pl 1.2-pl2 region as markers and DNA typing the resulting PCR amplification products to determine the presence of duplication and deletion in the corresponding chromosomal region, wherein Multiplex PCR amplification is carried out using 6 loci of D17S921, D17S9B, D17S9A, D17S918, D17S2230 and D17S4A as markers, and DNA-typing of the resulting PCR amplification products is carried out to determine duplication and deletion in the corresponding chromosomal region. In accordance with the method of the present invention, the diagnosis accuracy of detecting duplication and deletion in the chromosome 17pl 1.2-pl2 region is greater than 99.9%.

WO 2006/011716 A1

— *with sequence listing part of description published separately in electronic form and available upon request from the International Bureau*

For two-letter codes and other abbreviations, refer to the "Guidance Notes on Codes and Abbreviations" appearing at the beginning of each regular issue of the PCT Gazette.